Prenatal diagnoses of sex chromosome conditions

see also p 397

Parents need more than just accurate information

Women who receive a prenatal diagnosis of a chromosome abnormality remember the circumstances precisely. Years later, they recall the exact words used to deliver the news, and many regret the manner in which they were told.[1] They read between the lines that their fetus is no longer worthy of life and that their feelings about the pregnancy are not important.

A test result showing that a fetus has a chromosomal difference leaves women and their partners with a permanent and life-altering decision whether or not to continue the pregnancy, a decision they must be able to accept for the rest of their lives. What do women and their partners need to make such a decision? How can health care providers best help them?

Prenatal testing for chromosomal conditions has been offered in industrialized nations since the 1970s. Yet little research has been done on pretest counseling, the communication of abnormal results, their impact on parents' decision-making, or the long-term outcomes of such decisions.

Prenatal genetic counseling is provided by several different health care providers (see p 397), and if and how it is practiced is widely variable.[2] Prenatal testing (amniocentesis or chorionic villus sampling) is often performed without prenatal counseling, leaving women and their partners ill-prepared for an unexpected finding. Guidelines for prenatal testing have been issued by the American College of Obstetrics and Gynecology, but no practice standards exist in the United States for prenatal testing education and counseling.[3]

Abramsky et al performed a pilot study on the way that news is delivered to parents when prenatal testing reveals a sex chromosome abnormality in a fetus.[4] They studied some of the most ambiguous categories of prenatal diagnosis: health care providers know little about sex chromosome abnormalities, the literature is often out-of-date and conflicting, and women and their partners often choose not to terminate pregnancies.[5,6] The authors observed that parents often received little or inaccurate information.[2] The authors call for adherence to published guidelines, yet do not elaborate on the important additional needs of women and their partners during such crises.

Accurate descriptions of sex chromosome differences are critical. The decisions may be regrettable and the long-term outcomes devastating if a decision to terminate pregnancy is based on the misinformation, for example, that

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any of the conditions associated with sex chromosome abnormalities is comparable to Down syndrome.[4] Physicians are obligated to obtain useful, up-to-date information and to ensure that parents have an adequate opportunity to consider their decision with the help of an experienced health care provider, preferably someone well versed in medical genetics.

Even with accurate information about a chromosomal finding, there are many challenges. Although a diagnosis of Down syndrome (trisomy 21) or a sex chromosome abnormality (Turner or Klinefelter syndrome) may be relatively certain, questions remain about how the child will be affected. A child with Down syndrome born with a severe cardiac abnormality has a different prognosis from a child whose heart is unaffected. A girl with Turner syndrome who is short but otherwise appears normal may have a different life experience from a girl who is teased relentlessly for her appearance and correlates the social stigmatization with her feelings of self-worth. Health care providers can offer descriptions of populations of affected individuals, but no crystal ball exists for that particular fetus. In a recent study, parents of children with a rare condition, holoprosencephaly, consistently wanted health care providers to be honest about what they do not know and cannot predict about the ultimate outcome of an affected pregnancy.[7]

A 37-year-old woman undergoes amniocentesis because of advanced maternal age. She and her husband are conflicted about what they will do if the results indicate that the developing baby has Down syndrome. They concur that they want to know if the fetus is affected.

The results of the testing indicate that the fetus is a girl with an extra X chromosome, 47,XXX. The evidence suggests that she has an increased risk in childhood for speech delay, learning difficulties, and behavior problems.[8,9] Her physical appearance is unlikely to be affected, but there is a chance she could have a horseshoe kidney, patent ductus arteriosus, and bilateral hip dislocation.

Already ambiguous about how they would react to a diagnosis of Down syndrome, this unexpected news leaves them even more uncertain. The couple meets with a genetic counselor to discuss their family plans, support resources, and expectations for their daughter. Ultimately, they decide that they are willing to raise a child with 47,XXX, particularly because in most affected girls, the condition is never diagnosed. The parents remark, however, that the pregnancy has been spoiled for them and they wish that they didn't know this information about their daughter. They are concerned about how they will raise her without assuming that her chromosome status is responsible when she encounters problems.

Patients differ in the information they need about the condition. How that information is best imparted and how women and their partners use the information when making a decision are also important to understand. Reproductive decisions are complex and multifaceted, and information is only 1 component in the decision-making process.[10]

Although little empirical data show how reproductive decisions are made, clinical practice and theoretical models of other health care-related decisions suggest that they are influenced by women's values and beliefs, as well as their hopes and dreams for their children and family.[1,10,11] In particular, attitudes toward abortion, desires for biologic children, religious beliefs, attitudes toward disability and human variation, and social norms about prenatal testing outcomes are likely influences. So, too, are practical issues, such as money and social support. Thus, to help with decision-making, health care providers have an obligation to explore the meaning that the information has for women and their partners. If this counsel is not within the physicians' expertise, they should refer patients to a genetic counselor, nurse, or medical geneticist who can facilitate such decisions.

One of the hardest challenges in helping parents is the crisis that typically follows a prenatal diagnosis. If the woman and her partner are unprepared for the possibility, they may react strongly and 'shut down' emotionally. It is all but impossible to engage such patients in a decision-making process. Research involving individuals in crises suggests that physicians should not attempt to challenge defense mechanisms, but rather to work within them.[12] Individuals under stress may become hypervigilant, making rapid and ill-considered (and later regretted) decisions as an escape from the psychological distress.[13] Because typical counseling interventions are less likely to be effective, health care professionals should have experience facilitating decisions during crises.

Patients suffer a loss when they receive a prenatal diagnosis about their fetus. The loss is often not of the fetus they actually carry but of the fetus the parents had hoped they carried. This grief is profound but does not preclude a woman's ability to welcome an affected fetus into the world. Others, including health care professionals, may interpret this grief as rejection of the affected fetus. Women have been articulate about their resentment of such assumptions.[1] Often, the grief represents a readjustment of expectations. Women who experience this loss

appreciate those who accept it as part of adjustment and do not conclude from it an unwillingness to continue the pregnancy.

All women who receive a prenatal diagnosis suffer loss, regardless of their ultimate decision. During this crisis, they deserve not only accurate information, but also health care providers who convey respect, honesty, and compassion rather than those who provide swift answers or ill-informed suggestions about whether or not to continue the pregnancy.

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